CLAIMS

1. A method of diagnosing or monitoring a lysosomal storage disorder in a patient, comprising: measuring the level of at least one saposin in a tissue sample of the patient, wherein the level is an indicator of presence or extent of the disorder in the patient.

2. The method of claim 1, wherein the sample is a plasma sample.

3. The method of claim 1, wherein the sample is a whole blood sample.

4. The method of claim 1, wherein the measured level exceeds a mean level in a control population of individuals not having a lysosomal storage disorder, to indicate presence of the disorder in the patient.

5. The method of claim 1, further comprising measuring the level of the at least one saposin in a second tissue sample from the patient, the first and second samples being obtained at different times; and comparing the levels in the samples to indicate progression of the disease.

6. The method of claim 1, wherein the patient is undergoing treatment for the lysosomal storage disorder.

7. The method of claim 4, wherein the measured level is greater than the 95% percentile level in the control population.

8. The method of claim 1, wherein the patient is not known to have a lysosomal storage disorder before the measuring step.

9. The method of claim 1, wherein the patient is an infant less than one year old.

10. The method of claim 1, wherein the patient is a foetus and the sample is a foetal blood sample.

11. The method of claim 1, wherein the patient is known to have a lysosomal storage disorder and the level of the saposin indicates progression of the disorder.

12. The method of claim 1, wherein the patient is known to have a lysosomal storage disorder, and is being treated for the disorder, and the level of the saposin indicates response to treatment.

13. The method of claim 1, wherein the saposin is selected from the group consisting of saposin A, B, C, D, prosaposin, and mRNA encoding prosaposin.

14. The method of claim 1, wherein the saposin is selected from the group consisting of saposin A, C or D.

15. The method of claim 1, wherein the measuring step comprising detecting binding between a saposin polypeptide and an antibody.

16. The method of claim 15, wherein the antibody is a monoclonal antibody.

17. The method of claim 15, wherein the antibody is immobilised to a solid phase.

18. The method of claim 1, wherein the lysosomal storage order is selected from the group consisting of cystinosis, Fabry's disease, Niemann-Pick disease, Pompe's disease, and Wolman disease.

19. The method of claim 1, further comprising informing the patient or a parent or guardian thereof of the presence of the lysosomal storage disorder.

20. The method of claim 1, further comprising determining a treatment program based on the measurement.

21. A method of diagnosing or monitoring a lysosomal storage disorder in a patient, comprising: measuring the level of α-glucosidase in a tissue sample from a patient, wherein the level is an indicator of the presence or extent of the disorder in the patient.

22. The method of claim 21, wherein the sample is a plasma sample.

23. The method of claim 21, wherein the sample is a blood sample.

24. The method of claim 21, further comprising diagnosing the presence of a disorder selected from the group consisting of acid lipase disease, mannosidosis, MPSII, MPS IIIA, MSD, mucolipidosis, N-P (A/B), N-P (C), Sandhoff, SAS or TSD B1, if the measured level of α-glucosidase exceeds the mean level in a control population of individuals not having a lysosomal storage disease.

25. The method of claim 21, further comprising diagnosing the presence of disorder selected from the group consisting of galactosialidosis, MPS IVA and Pompe's disease if the measured level of α-glucosidase is below the mean level in a control population of individuals not having lysosomal storage disease.

26. A method of diagnosing a lysosomal storage disorder comprising measuring a level of a saposin in a tissue sample from the patient; measuring a level of LAMP-1 or LAMP-2 in a second tissue sample from the patient; measuring a level of α glucosidase in a third tissue sample from the patient; wherein an increased level of saposin and/or LAMP-1 or LAMP-2, and/or an increased or decreased level of α-glucosidase in the sample relative to respective mean levels in a control population is an indicator of presence or extent of the disorder in the patient.

27. A method of diagnosing Pompe's disease in a patient, comprising measuring a level of a saposin in a tissue sample from the patient: measuring the level of α-glucosidase in a second tissue sample from the patient; wherein the presence of an increased level of the saposin and a decreased level of the α-glucosidase relative to mean levels of the saposin and α-glucosidase in a control population of individuals not having a lysosomal storage disorder indicates Pompe's disease or susceptibility thereto.

28. A method of screening patients for presence of lysosomal storage disorder, comprising: measuring the level of a LAMP-1 polypeptide in a sample from the patient: measuring the level of a saposin peptide in the sample, the presence of an increased level of LAMP-1 or saposin or both relative to mean levels in a control population, indicating susceptibility to a lysosomal disorder.

29. A diagnostic kit comprising: a first reagent that binds to a LAMP; a second reagent that binds to a saposin.

30. The diagnostic kit of claim 29 , further comprising a third reagent that binds to α glucosidase.

31. The diagnostic kit of claim 30, wherein the first, second and third reagents are antibodies.

32. In a method of screening a patient for presence or susceptibility to disease, comprising performing a plurality of diagnostic tests on a tissue sample from the patient for a plurality of diseases, the improvement wherein one of the diagnostic tests comprises measuring the level of a saposin.

33. In the method of claim 32, the further improvement wherein a second of the diagnostic tests comprising measuring the level of LAMP-1 in the tissue sample from the patient.

34. In the method of claim 33, the further improvement wherein a third of the diagnostic tests comprises measuring the level of α-glucosidase in the tissue sample from the patient.

35. In the method of claim 32, the further improvement wherein a fourth of the diagnostic test comprises analysing a nucleic acid encoding an enzyme associated with a lysosomal storage disorder for a polymorphic form correlated with the disorder.

36. A method of monitoring treatment of a lysosomal storage disease in a patient, comprising determining a baseline level of a saposin in a tissue sample from the patient with a lysosomal storage disorder before treatment with an agent; comparing a level of the saposin in a tissue sample from the patient obtained after treatment with the agent; wherein a reduction in the level after treatment relative to the baseline indicates a positive treatment outcome.

37. A method of monitoring treatment of acid lipase disease, mannosidosis, MPSII, MPS IIIA, MSD, mucolipidosis, N-P (A/B), N-P (C), Sandhoff, SAS or TSD B1, comprising: determining a baseline level of α glucosidase in a tissue sample from the patient with the disorder before treatment with an agent; comparing a level of the α glucosidase in a tissue sample from the patient with the disorder after treatment with the agent with the baseline level; wherein a decrease relative to the baseline indicates a positive treatment outcome.

38. A method of monitoring a patient with Pompe's disease, comprising: determining a baseline level of α glucosidase in a tissue sample from the patient with the disorder before treatment with the agent; comparing a level of the α-glucosidase in a tissue sample from the patient after treatment with the agent with the baseline level; wherein an increase relative to the baseline indicates a positive treatment outcome.